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Letter to the Editor

Clinical and Molecular Studies in Full Trisomy 22: Further Delineation of the Phenotype and Review of the Literature. Reply to Dr. Hirschhorn

To the Editor:

We read with interest the comments by Dr. Hirschhorn regarding our recent paper on trisomy 22 [Bacino et al., 1995].

Dr. Hirschhorn refers to a case of a term newborn infant with multiple congenital anomalies who, on chromosome analysis, had full trisomy 22 [Hirschhorn et al., 1973]. We were aware of this report, but in the absence of more clinical details and patient photographs we did not include it in our paper. There was no description of ear malformations, webbing of the neck, digital anomalies, and the cardiac abnormality described did not appear to be a conotruncal defect. As it was stated in our paper we think that the main findings of this phenotype include: growth retardation, microcephaly, hypertelorism, microtia, webbed neck, conotruncal anomalies, digital defects, renal/genitourinary anomalies and anorectal abnormalities. On the other hand, Hirschhorn et al. [1973] describe a number of anomalies that are indeed present in many of the patients reviewed and seen by us. It will be very important to bring this case to the literature again if more clinical information is still available. Our selection of cases from the literature includes the ones who were confirmed by molecular methods and the ones that clinically share common elements with that group, even in the absence of molecular data. These criteria were used as an attempt to delineate more clearly the phenotype and do not reflect all cases of trisomy 22 that have been described.

REFERENCES

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